



## KRT83 gene

keratin 83

### Normal Function

The *KRT83* gene provides instructions for making the type II hair keratin K83 protein (K83). This protein belongs to a group of proteins known as keratins, which are tough, fibrous proteins that form the structural framework of cells that make up the hair, skin, and nails. Each keratin protein partners with another keratin protein to form molecules called intermediate filaments. These filaments assemble into strong networks that provide strength and resiliency to the tissues and protect them from being damaged by everyday physical stresses. The K83 protein is found in cells that make up the inner compartment of the hair shaft known as the cortex, and this protein helps give hair its strength and elasticity.

### Health Conditions Related to Genetic Changes

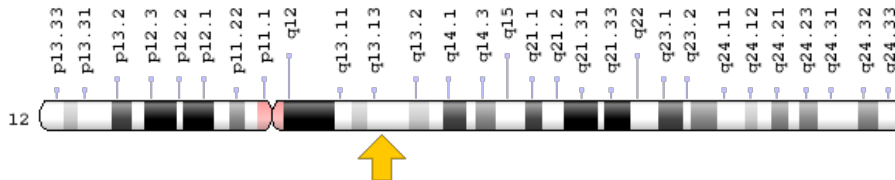
#### monilethrix

Mutations in the *KRT83* gene can cause monilethrix, a hair condition characterized by strands of hair with a beaded appearance and short, brittle hair that breaks easily. Mutations associated with this condition change a single protein building block (amino acid) in the K83 protein. The amino acid changes usually occur in a region of the K83 protein thought to be important in intermediate filament formation. In people with monilethrix, the cortex of the affected hair shaft appears abnormal. However, it is unclear how mutations in the *KRT83* gene are related to the abnormality in the cortex or the beaded appearance of the hair.

## Chromosomal Location

Cytogenetic Location: 12q13.13, which is the long (q) arm of chromosome 12 at position 13.13

Molecular Location: base pairs 52,314,301 to 52,321,398 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- hair keratin K2.10
- hard keratin, type II, 3
- Hb-3
- HB3
- hHb3
- K83
- keratin-83
- keratin 83, type II
- keratin, hair, basic, 3
- KRT83\_HUMAN
- KRTHB3
- type II hair keratin Hb3
- type-II keratin Kb23

## Additional Information & Resources

### Educational Resources

- Madame Curie Bioscience Database: Development and Structure of the Hair Follicle  
<https://www.ncbi.nlm.nih.gov/books/NBK45997/#ch4829.s2>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28KRT83%5BTIAB%5D%29+OR+%28%28hHb3%5BTIAB%5D%29+OR+%28K83%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- KERATIN 83, TYPE II  
<http://omim.org/entry/602765>

### Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=KRT83%5Bgene%5D>
- HGNC Gene Family: Keratins, type II  
<http://www.genenames.org/cgi-bin/genefamilies/set/609>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=6460](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6460)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/3889>
- UniProt  
<http://www.uniprot.org/uniprot/P78385>

### **Sources for This Summary**

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